V. Genetics Home Reference

http://ghr.nlm.nih.gov

The Genetics Home Reference is the National Library of Medicine's web site for consumer information about genetic conditions and the genes responsible for those conditions. The language is written at the high-school level -- for those who remember some of their high school biology course.

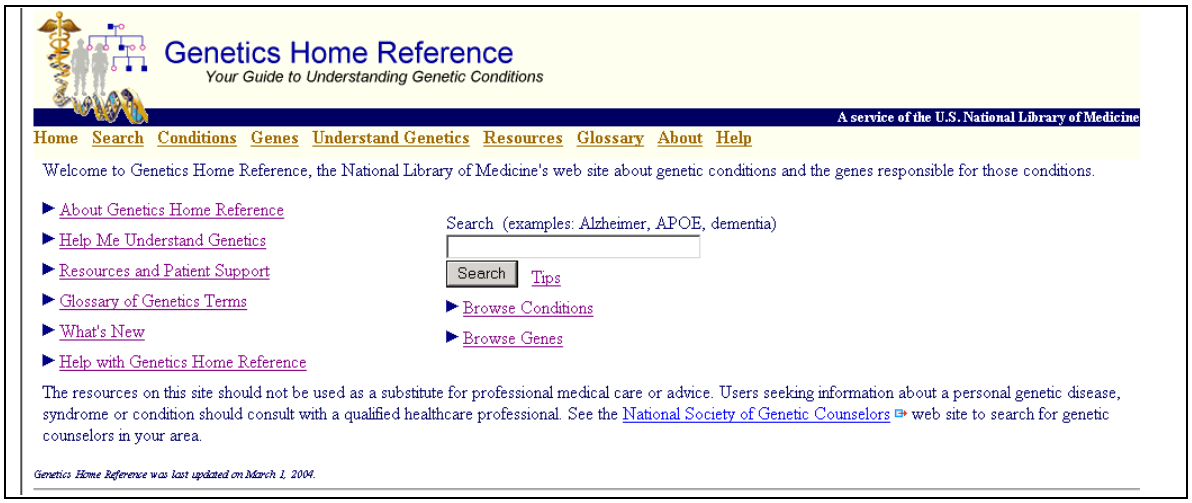
Genetics Home Reference contains:

- Genetic Condition Summaries
  - Each summary explains the condition's genetic cause and pattern of inheritance.
- Gene Summaries
  - Each summary provides the official name and symbol of a gene, its chromosomal location, and an explanation of its normal function and how mutations in the gene cause particular genetic conditions.
- Help Me Understand Genetics
  - This document provides a basic explanation of how genes work and how mutations cause disorders. It also includes current information about genetic testing, gene therapy, and the Human Genome Project.
- Glossary
  - A searchable glossary to find definitions of genetic and medical terms is available from every web page. In addition, each summary provides a list of glossary terms used on the page, with a direct link to their definitions.

The health topics selected for GHR are the most common ones in MedlinePlus that have a genetic component. At this point only conditions caused by mutations in genes are included. Conditions caused by duplicated, missing, or rearranged chromosomes, will be summarized at a later date.

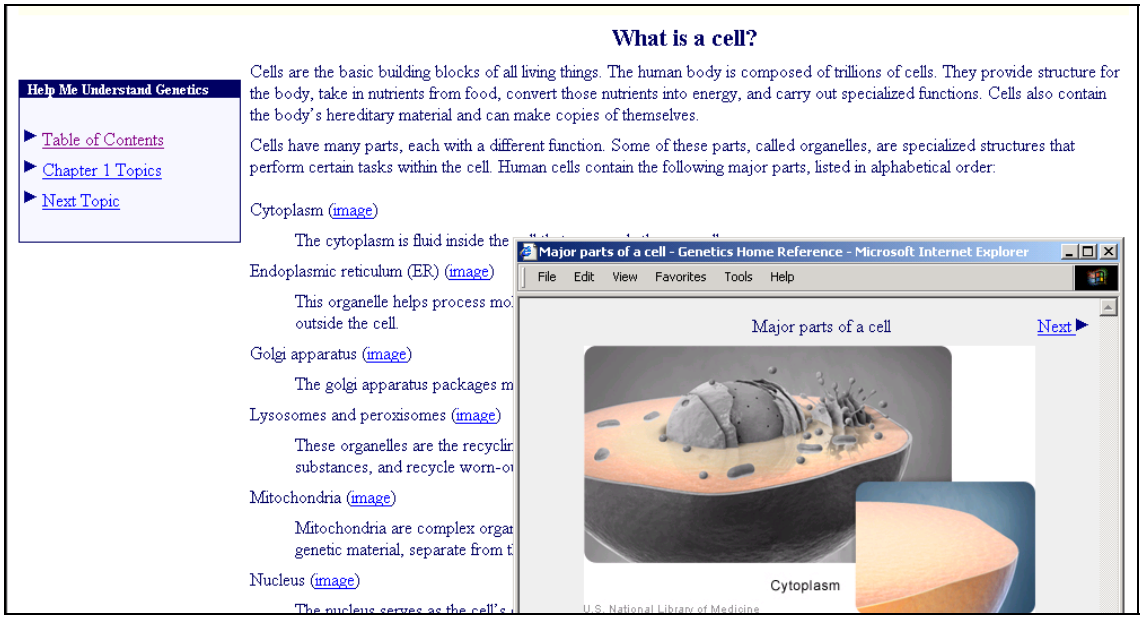
Genetics Home Reference is a service of the U.S. National Library of Medicine, part of the National Institutes of Health

Note that *on the home page*, the menu in the header is somewhat the same as the menu on the left and center of the page. On subsequent pages, the left menu changes, while the menu in the header does not.



Help Me Understand Genetics

Written for the layperson, *Help Me Understand Genetics* is a small online "book" that carefully explains genetics starting with "What is a Cell?" Excellent linked illustrations make this book even easier to understand. The illustrations open in a small separate window so you can read and look at the illustrations at the same time.



There is even a printable version of the book, which includes the illustrations within the text.

Searching Genetics Home Reference

Genetic Conditions and Genes can be searched two ways. They can be browsed or searched by keyword.

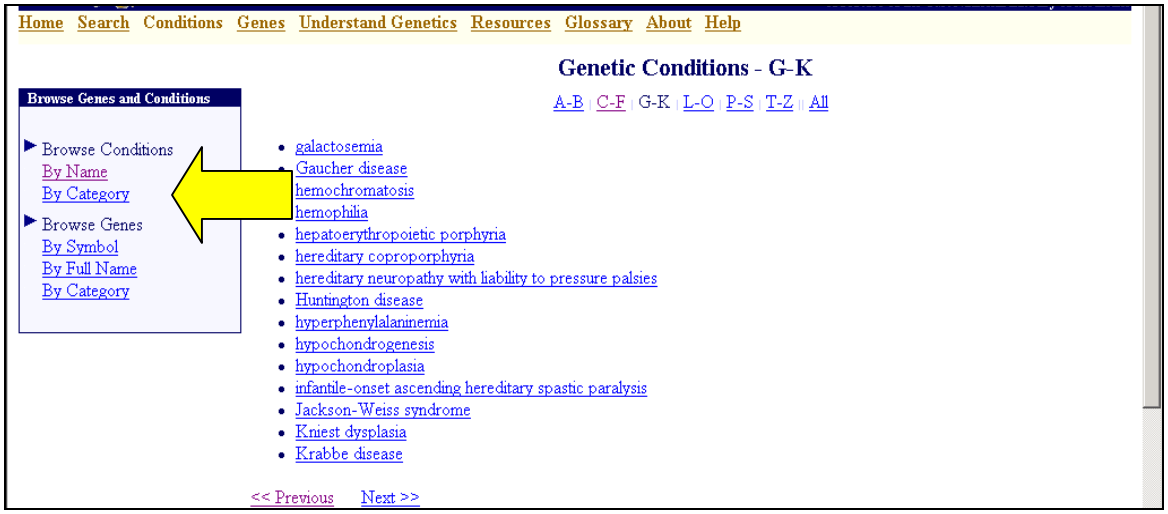
Keyword search:

Keyword search is probably best if you aren't sure what you are looking for, or if you want as much information as possible.

- You can type any word or phrase that is relevant to your search in the box in the center of the page. For example "forgetfulness" will take you to the page that has that term in its text, in this case "Alzheimer Disease."
- When using Boolean terms in the search, be sure to use uppercase letters.
- If you want a search to contain an exact match to a word (e.g. to match "balding," but not "bald" or "baldness), use the "plus" symbol: +balding.

Browse search:

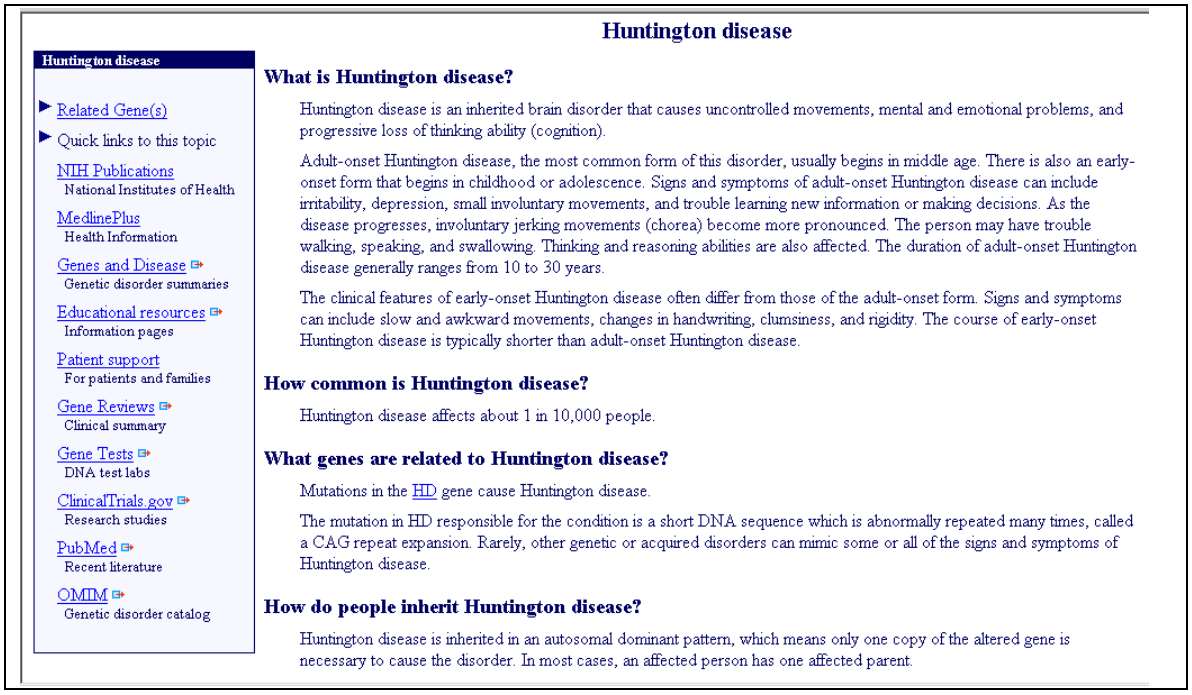
The browse search can be used for both Genes and Conditions. Browsing can be done by Name, Category, or (in the case of Genes) Symbol.



Search Results

Genetic Conditions

Clicking on a particular genetic condition will link you to a page with a great deal of information



Huntington disease

Huntington disease

- Related Gene(s)
- Quick links to this topic

NIH Publications
National Institutes of Health

MedlinePlus
Health Information

Genes and Disease
Genetic disorder summaries

Educational resources
Information pages

Patient support
For patients and families

Gene Reviews
Clinical summary

Gene Tests
DNA test labs

ClinicalTrials.gov
Research studies

PubMed
Recent literature

OMIM
Genetic disorder catalog

What is Huntington disease?

Huntington disease is an inherited brain disorder that causes uncontrolled movements, mental and emotional problems, and progressive loss of thinking ability (cognition).

Adult-onset Huntington disease, the most common form of this disorder, usually begins in middle age. There is also an early-onset form that begins in childhood or adolescence. Signs and symptoms of adult-onset Huntington disease can include irritability, depression, small involuntary movements, and trouble learning new information or making decisions. As the disease progresses, involuntary jerking movements (chorea) become more pronounced. The person may have trouble walking, speaking, and swallowing. Thinking and reasoning abilities are also affected. The duration of adult-onset Huntington disease generally ranges from 10 to 30 years.

The clinical features of early-onset Huntington disease often differ from those of the adult-onset form. Signs and symptoms can include slow and awkward movements, changes in handwriting, clumsiness, and rigidity. The course of early-onset Huntington disease is typically shorter than adult-onset Huntington disease.

How common is Huntington disease?

Huntington disease affects about 1 in 10,000 people.

What genes are related to Huntington disease?

Mutations in the HD gene cause Huntington disease.

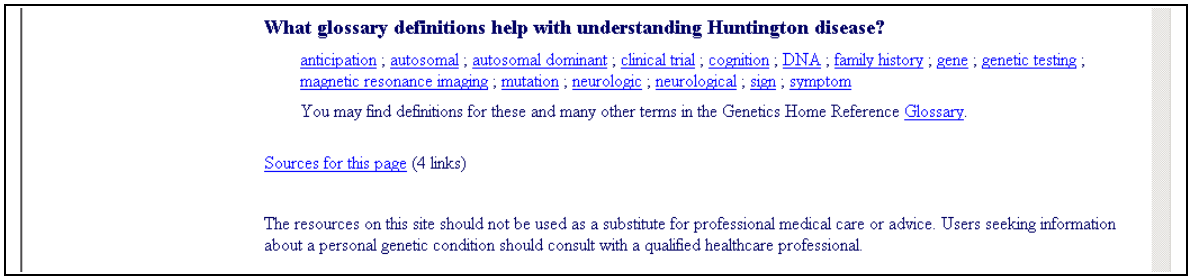
The mutation in HD responsible for the condition is a short DNA sequence which is abnormally repeated many times, called a CAG repeat expansion. Rarely, other genetic or acquired disorders can mimic some or all of the signs and symptoms of Huntington disease.

How do people inherit Huntington disease?

Huntington disease is inherited in an autosomal dominant pattern, which means only one copy of the altered gene is necessary to cause the disorder. In most cases, an affected person has one affected parent.

Quick links on the left lead you to established websites in a variety of locations.

At the bottom of the page are links to the glossary on specific words that will help with understanding the particular condition that is being researched.



What glossary definitions help with understanding Huntington disease?

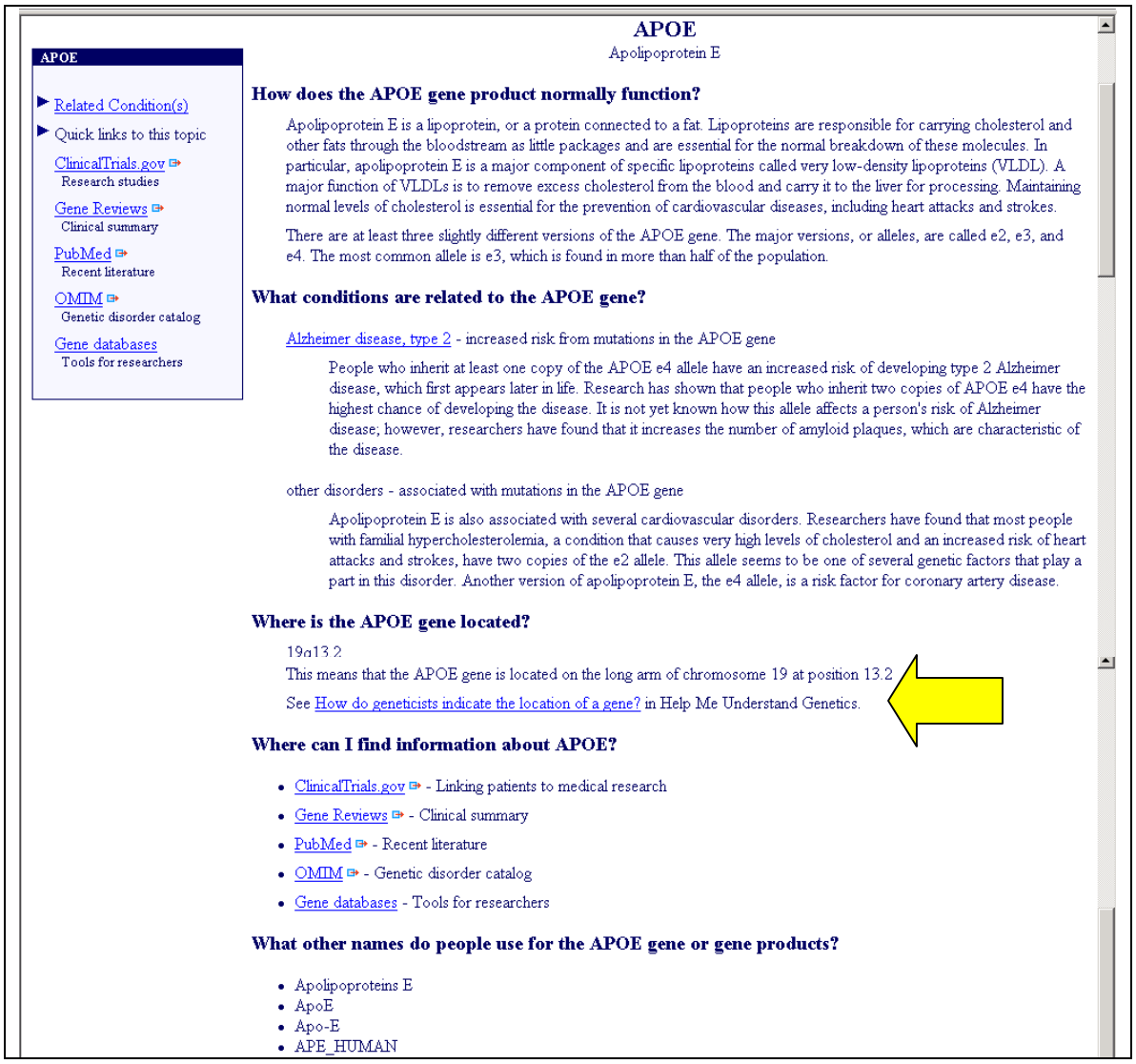
anticipation ; autosomal ; autosomal dominant ; clinical trial ; cognition ; DNA ; family history ; gene ; genetic testing ; magnetic resonance imaging ; mutation ; neurologic ; neurological ; sign ; symptom

You may find definitions for these and many other terms in the Genetics Home Reference Glossary.

Sources for this page (4 links)

The resources on this site should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic condition should consult with a qualified healthcare professional.

Genes



APOE

Apolipoprotein E

- Related Condition(s)
- Quick links to this topic

ClinicalTrials.gov
Research studies

Gene Reviews
Clinical summary

PubMed
Recent literature

OMIM
Genetic disorder catalog

Gene databases
Tools for researchers

How does the APOE gene product normally function?

Apolipoprotein E is a lipoprotein, or a protein connected to a fat. Lipoproteins are responsible for carrying cholesterol and other fats through the bloodstream as little packages and are essential for the normal breakdown of these molecules. In particular, apolipoprotein E is a major component of specific lipoproteins called very low-density lipoproteins (VLDL). A major function of VLDLs is to remove excess cholesterol from the blood and carry it to the liver for processing. Maintaining normal levels of cholesterol is essential for the prevention of cardiovascular diseases, including heart attacks and strokes.

There are at least three slightly different versions of the APOE gene. The major versions, or alleles, are called e2, e3, and e4. The most common allele is e3, which is found in more than half of the population.

What conditions are related to the APOE gene?

Alzheimer disease, type 2 - increased risk from mutations in the APOE gene

People who inherit at least one copy of the APOE e4 allele have an increased risk of developing type 2 Alzheimer disease, which first appears later in life. Research has shown that people who inherit two copies of APOE e4 have the highest chance of developing the disease. It is not yet known how this allele affects a person's risk of Alzheimer disease; however, researchers have found that it increases the number of amyloid plaques, which are characteristic of the disease.

other disorders - associated with mutations in the APOE gene

Apolipoprotein E is also associated with several cardiovascular disorders. Researchers have found that most people with familial hypercholesterolemia, a condition that causes very high levels of cholesterol and an increased risk of heart attacks and strokes, have two copies of the e2 allele. This allele seems to be one of several genetic factors that play a part in this disorder. Another version of apolipoprotein E, the e4 allele, is a risk factor for coronary artery disease.

Where is the APOE gene located?

19q13.2

This means that the APOE gene is located on the long arm of chromosome 19 at position 13.2

See How do geneticists indicate the location of a gene? in Help Me Understand Genetics.

Where can I find information about APOE?

- ClinicalTrials.gov - Linking patients to medical research
- Gene Reviews - Clinical summary
- PubMed - Recent literature
- OMIM - Genetic disorder catalog
- Gene databases - Tools for researchers

What other names do people use for the APOE gene or gene products?

- Apolipoproteins E
- ApoE
- Apo-E
- APE_HUMAN

Note that when explanations require knowledge of genetics, the links refer you to the chapter in *Help Me Understand Genetics* that will explain that aspect of genetics.

Resources and Patient Support

Resources include:

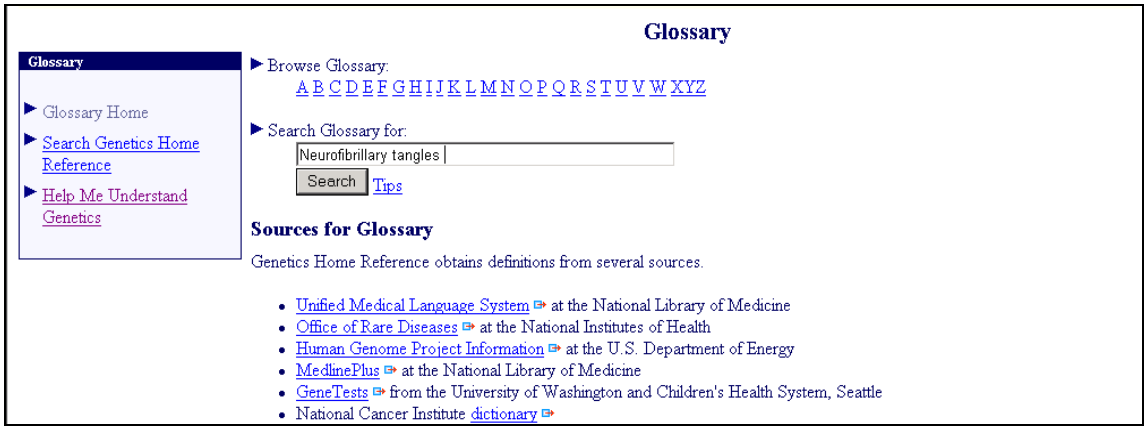
- News from the CDC, Genome News Network, and MedlinePlus
- Educational sites about genetics from government and educational agencies.
- Sites specifically about the Human Genome Project
- Medical and Genetic Data resources for health professionals and researchers.

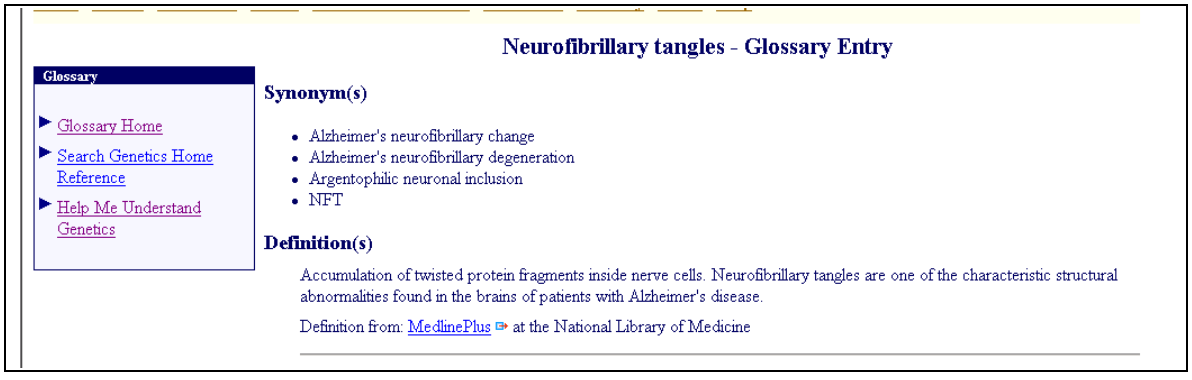
The Patient Support section has links to

- Genetic Alliance, which has a tool for creating a family history
- Children and Adolescents with Special Care Needs in Maternal and Child Health Library
- The National Organization for Rare Disorders, which has a free index of rare diseases with general descriptions, and support organizations.
- Links to support organizations by genetic condition.

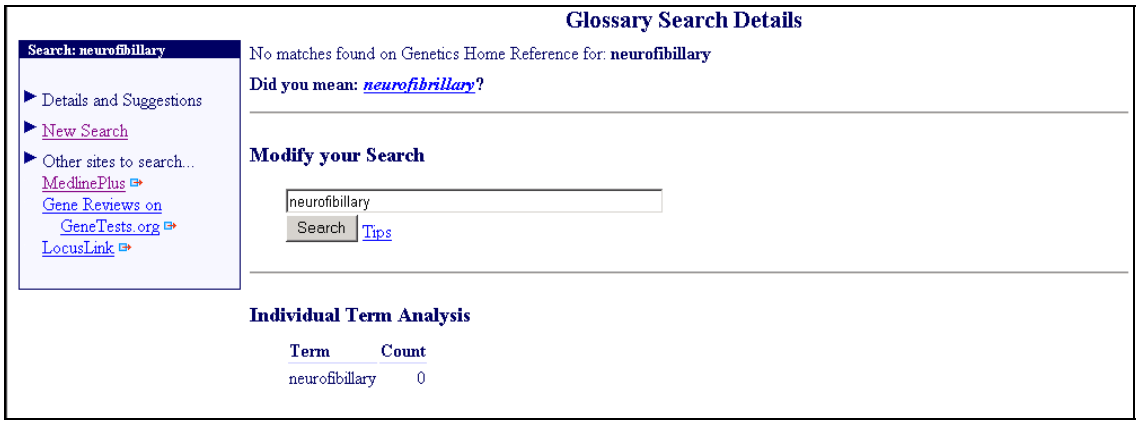
Glossary

The glossary has simple definitions of words found in Genetics Home Reference. The sources for the definitions are found at the bottom of the page.





If you spell the word incorrectly, the glossary will attempt to find the correct word for you.



Sponsorship:

The Genetics Home Reference is a service of the U.S. National Library of Medicine, part of the National Institutes of Health, an agency of the Department of Health and Human Services. The project team includes board-certified medical geneticists, biologists, and computer and information scientists.

Practice Questions

1. How do you get a genetic mutation? Do they come from the sun's radiation, like in Mutant X?

2. Can you inherit Lou Gehrig's disease?

3. I need to know the history of the study of genetics to back up my science project on the Human Genome Project. Do you know where I can find something?